Rhode Island Department of Health
Genetic Screening Advisory Committee
Annual Report 2000

The Rhode Island Genetic Screening Advisory Committee was formed in September of 1999 and is currently chaired by the Director of Health, Patricia A. Nolan, MD, MPH. Members of the committee include consumers, ethicists, academicians, public health professionals, medical geneticists, and other healthcare providers. The Committee meets approximately once a month at the Rhode Island Department of Health (HEALTH). The current membership is listed on the HEALTH website at www.healthri.org

The mission of the RI Genetic Screening Advisory Committee is to advise HEALTH on decision making processes that could serve us over the years and would allow for a full discussion of the issues in genetics and public health. In support of this mission, the Committee explored criteria for selecting newborn screening tests in Rhode Island. This report documents the perspectives and recommendations at the end of the Committee's first year.

At the same time HEALTH staff worked to develop awareness and expertise to address the implications of the new genetics. Two statewide genetic conferences were held the first in January 2000 "Translating the Advancements of Genetics in Public Health for Rhode Island" and in November 2000 on "Exploration of Confidentiality in the New World of Genetics". Staff formulated a genetic strategic plan for HEALTH funded by the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA).

The Committee started with a focus on newborn screening tests, which are within the purview of HEALTH's authority. The Committee realized that to recommend a change in newborn screening tests all the other aspects of the newborn screening program would have to be addressed including funding, provider education, laboratory capacity, community support services, etc.

To meet the challenge, three subcommittees have been constituted, namely, the "Guidelines for Decision Process" Subcommittee for selecting newborn screening criteria, the "Newborn Screening Subcommittee", and the "Ethics" Subcommittee. These met periodically at the discretion of the

subcommittee chairs. The composition of the subcommittees is given on the HEALTH website. The subcommittee reports are summarized below.

Summary of Subcommittee on Guidelines for Decision Process

This Subcommittee was charged with defining the standards for HEALTH to use when evaluating new or existing newborn screening tests. The Subcommittee work was iterative in nature with extensive discussion at full committee meetings. The subcommittee concluded that 1) when evaluating whether or not to recommend a newborn screen, the final decision must be based on multiple factors, not a single factor, and 2) Support services must be in place prior to instituting an additional newborn screening, even when the test is offered on a voluntary basis.

The Guidelines for assessing newborn screening tests are as follow:

- Tests should be conducted for conditions severe enough to warrant early detection and to justify allocation of public health resources.

- There should be an early benefit of detection and timely intervention (before onset of irreparable harm). Benefit may be defined as one or more of the following:
  - Prevention of premature death
  - Prevention of disease
  - Improved function

- Universal Newborn Screening should be performed with tests of demonstrated validity (sensitivity, specificity, and predictive value).

- Infrastructure to care for the condition should adhere to national standards: e.g., resources should be adequate for diagnosis, genetic counseling and treatment.

- Issues such as confidentiality, parental consent, transportation, cultural and language diversity, professional education, family education and counseling should be considered and addressed.

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Summary of Subcommittee on Newborn Screening

The subcommittee activities over the past year have been focused on the subcommittee members' role as expert advisors on medical, epidemiological and clinical issues related to newborn screening. Advances in molecular genetics and advances drive this discussion on medical technology. The subcommittee felt it was very important to address each of these issues and advise the full committee as to the current status. Important decisions were at hand for the state of Rhode Island on whether or not to initiate screening in each of these areas.

Molecular Genetic Screening

The prototype for the ability of new technologies to provide molecular genetic screening is cystic fibrosis. The state of Wisconsin has been doing molecular genetic testing for cystic fibrosis as a newborn screen for the past several years. The state of Massachusetts initiated molecular genetic screening for cystic fibrosis based on a pre-symptomatic, newborn screening approach approximately one year ago. The subcommittee reviewed the medical literature on newborn screening for cystic fibrosis, as well as contacting members involved with the programs in Wisconsin and Massachusetts. The subcommittee positions on July 24, 2000 were as follows:

1. Cystic fibrosis can be diagnosed based on newborn screening.
2. The reliability of this diagnosis depends on the population being screened and the number of genetic forms of cystic fibrosis included in the test.
3. The screening test is not 100% sensitive, since there are several hundred genetic forms of cystic fibrosis making molecular analysis of all forms infeasible.
4. The evidence for the benefits of pre-symptomatic diagnosis of cystic fibrosis was reviewed and found inconclusive.
5. The subcommittee recommended that this is an important

contemporary issue that should be monitored closely (on a not less than quarterly basis) for available new information.

Tandem Mass Spectrometry Metabolic Screening

New technologies make it feasible to screen for important metabolic diseases. The prototype new technology is tandem mass spectrometry, also known as "MS/MS." The prototypic disease for which tandem mass spectrometry permits screening is called medium-chain acyl CoA dehydrogenase deficiency or MCAD. MS/MS screening for MCAD and other metabolic diseases is now taking place in Massachusetts, New Hampshire and Maine among the New England consortium and in nine other states in the United States. Additionally, there are a number of states that are initiating MS/MS screening programs. The subcommittee reviewed the literature on MS/MS screening for MCAD and other diseases. A formal presentation was made by the "NeoGen" Corporation, which is a private commercial enterprise providing newborn screening. It was felt that MS/MS screening was an important, exciting new technology likely to be beneficial but that the benefits of pre-symptomatic diagnosis of MCAD and the effectiveness of clinical interventions need additional review at this time. It was felt that this important issue should be monitored on not less than a quarterly basis and that screening for MCAD and/or other metabolic diseases by MS/MS may need to be adopted for Rhode Island in the near future.

Each of these issues has been re-visited by the subcommittee members individually and as a group. At its most recent meeting on 8/16/00 the subcommittee felt the available information to support screening for MCAD and other fatty acid oxidation defects had now shifted the balance toward recommending its adoption. In the next quarter, the subcommittee plans to collate and codify that data and will make a formal recommendation to the full committee.

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Summary of Subcommittee on Ethics

The Ethics Subcommittee met once on November 29, 1999 to respond to questions posed to it by the full committee; namely, a) what are the ethical issues in public health genetic screening? and b) what ethical principles ought to guide mandating, offering and avoiding government-supported genetic screening/testing? Potential concerns, harms, and benefits were identified as well as ethical principles that ought to guide the selection of genetic conditions for screening by the Rhode Island Department of Health.

There are potential concerns as well as benefits to genetic screening performed by a public health/government service. Potential concerns include:

- Discrimination/stigmatization
- Loss of privacy/confidentiality
- Costs including cost effectiveness fair distribution and potential liability.
- Unreasonable fears/expectations and conclusions
- Limits of validity
- Reasonable fears such as knowing one's "crystal ball"
- Knowledge without appropriate intervention
- Potential for stimulating "eugenic" attitudes/behaviors.

Potential benefits include:

- Proper use by the individual of available (and inevitable) knowledge and technology to assist in life planning. This creates a need for effective public education, where the responsibility would fall on the Department of Health
- The right to know (versus the need to know)
- Societal health planning

Ethical principles that ought to guide the selection of genetic screening:

- The risk/benefit ratio should show that the benefits outweigh the harm to both the individual and to society.
- Sufficient numbers of individuals will be involved to justify the effort and expense which responds to the justice/fairness issue.
- The information will be understandable and appropriately used by the public when effective interventions become available.

More specific issues were considered:

- The ethical issues vary with the kind of screening under consideration. The issues appear to have critical differences and emphases when dealing with prenatal, neonatal, children and adult screening.
- What other states do concerning genetic screening and their rationale behind it may have relevance for Rhode Island
- Identifying and discussing conditions/diseases that meet the above criteria would serve as a model for future deliberations.

Future Actions

The Director of Health will ask the Advisory Committee for direction on the course of action for the near future, which includes:

- ✓ Establish a process to add newborn-screening tests to the eight currently authorized in Rhode Island. Would the recommended test be mandatory, voluntary, research and/or a pilot? Is the statutory authority sufficient to add a test through rule making? Identify the funding including insurance coverage issues and how that would fit with the current funding mechanisms. Identify the community and HEALTH responsibilities for adding a new test.
- ✓ Guidance for consent to test that incorporates the various types of tests recommended.
- ✓ Responsibility for follow-up when a positive test result is reported. What are the standards for capacity, training, communication, etc. required prior to approving another test?
- ✓ Address issues of multiple vendors.
- ✓ Should the committee reorganize itself and form another subcommittee to address these issues?
- ✓ How to interface with the Legislative Commission and genetic related 2001 legislation.
- ✓ Broaden the mission of this interdisciplinary committee to advise on genetic testing for the whole population.

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The Genetic Screening Advisory Committee will continue its work and expects to recruit new participants whose expertise is pertinent for the next topics for consideration.

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